MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
TESTINGRNV,CACTK	SA00047499	27	M	SA00047499

Ordering Phys		DOB
		06/01/1985

Client Order #	Account Information	Report Notes
SA00047499	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE ROCHESTER,MN 55901	
Collected 07/09/2012		
Printed 08/02/2012 15:50		

Test	Flag	Results	Unit	Reference Value	Perform Site*
SLC25A20 Gene, Known Mutation		REPORTED 07/11/2012 15:00			
Specimen		Blood			MCR
Specimen ID		1038356			MCR
Order Date		11 Jul 2012 14:56			MCR
Reason For Referral					MCR
Family history of carnitine acyl-carnitine translocase (CACT) deficiency. Test for the presence of mutations in the SLC25A20 gene.					
Method					MCR
DNA sequence analysis was used to test for the presence of the c. 362delG alteration in exon 4 of the SLC25A20 gene (GenBank accession number; NM_000387.5). Analysis for this specific alteration was performed because it has been identified in a family member.					
Result					MCR
The c.362delG alteration was NOT detected.					
Interpretation					MCR
Absence of the mutation previously identified in an affected family member indicates that this individual is at no greater risk than someone in the general population for developing symptoms related to CACT deficiency.					
This assay does not rule out the presence of other disease causing mutations in this gene or other genes that are associated with metabolic disease. Errors in the diagnosis or pedigree provided to us, including non paternity, may lead to an erroneous interpretation of test results.					
A genetic consultation may be of benefit.					
A list of common polymorphisms identified for this patient is available upon request.					
CAUTIONS: Test results should be interpreted in context of clinical findings, family history, and other laboratory data. Misinterpretation of results may occur if the information provided is inaccurate or incomplete.					
Rare polymorphisms exist that could lead to false negative or positive results. If results obtained do not match the clinical findings, additional testing should be considered.					

Performing Site Legend on Last Page of Report

* Report times for Mayo performed tests are CST/CDT

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Test	Flag	Results	Unit	Reference Value	Perform Site*
Bone marrow transplants from allogenic donors will interfere with testing. Call Mayo Medical Laboratories for instructions for testing patients who have received a bone marrow transplant.					
Laboratory developed test.					
Reviewed By					MCR
Melody Elizabeth Kimball					
Release Date		11 Jul 2012 14:59			MCR

* Performing Site:

MCR	Mayo Clinic Laboratories - Rochester Main Campus 200 First St SW Rochester, MN 55905	Lab Director: Franklin R. Cockerill, III, M.D.

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